Median facial dysplasia: A review

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ABSTRACT

Three cases of Median facial dysplasia are presented with their classification, embryological basis and management strategy.

KEY WORDS

Median facial dysplasia, Holoprosencephaly.

INTRODUCTION

Birth defects of the face are often the most disturbing because we usually identify and differentiate people by the appearance of their face. Noordhoff and Cheng (1982) described a discrete subgroup in cleft lip and palate patients.¹ This group called as Median facial dysplasia (MFD) which exhibits certain characteristic craniofacial defects like lack of Cupid’s bow, short prolabium, absence of frenulum labii, hypoplasia of premaxilla, absent upper central and lateral incisors on cleft side, deficient septal cartilage and nasal spine. Gross brain abnormalities are usually absent in MFD. The same facial malformations are also described in patients with holoprosencephaly sequence (HPE-S) but they are associated with gross abnormalities of the brain in lobar and semilobar holoprosencephaly.² This can be explained by the similar embryological origin of MFD and HPE-S. Median facial dysplasia should be distinguished from the mid face hypoplasia which includes the underdevelopment or posterior positioning of the inferior portion of the orbits, nasal bone and maxilla. Mid face hypoplasia can be a part of MFD but the reverse is not true. The diagnosis of median facial dysplasia is made in the presence of midfacial deficiencies in a unilateral or bilateral cleft lip with or without cleft palate and without clinically detectable anomalies of the brain. This group of patients consists of only 2% of cleft lip and palate patients.¹ These patients have normal intelligence level but have inherent potential for poor mid face growth. Therefore, it is important to identify this subgroup of patients as they have serious growth problems and pose difficulty in reconstruction. Early recognition of this entity is important for counseling parents and several alterations in standard operative methods and orthodontic protocols are necessary. It is also important to determine the degree of holoprosencephaly since psychomotor development and survival depends upon it, which in turn will influence the management plan.

CASE REPORTS

Three male patients with false median cleft lip without hypotelorism and without holoprosencephaly were seen during last two years. On examination all had median cleft lip with rudimentary prolabium, poorly defined or absent cupids bow, absent upper labial frenulum, flattened nose with widening of nostril,
absent nasal spine, deficiency of columella, nasal septal cartilage and vomer. Premaxilla was absent in one patient and rudimentary in the remaining two patients. All were missing one maxillary incisor. (Figures 1 to 3 and Table 1) One patient had midline cleft of secondary palate and one had multiple enchondromas over extremities. Two patients had developmental delays. CT scan head of patient No. 2 and 3 was normal. Dental occlusion in patient number 3 was normal while patient 2 had class three malocclusion. Dental occlusion in patient 1 could not be ascertained as the teeth were not erupted yet.

All the patients were full term normal delivery. Pregnancy was uncomplicated without any history of teratogenic exposure. No family history of craniofacial malformation was found on either side. The diagnosis was made in the presence of typical findings of deficient midline facial structures and absence of gross brain abnormalities. Surgical correction of lip was performed by using small prolabium for lip reconstruction in all patients. (Figure 4)

Table 1: Showing clinical findings in patients with median facial dysplasia

<table>
<thead>
<tr>
<th>S. No.</th>
<th>Age</th>
<th>Development</th>
<th>C T scan brain</th>
<th>Associated anomalies</th>
<th>Operations</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patient1</td>
<td>10 months</td>
<td>Delayed</td>
<td>Not done</td>
<td>Mid line cleft palate</td>
<td>Lip repair</td>
</tr>
<tr>
<td>Patient2</td>
<td>12 years</td>
<td>Mild delay</td>
<td>Normal</td>
<td>Multiple enchondromas</td>
<td>Lip repair</td>
</tr>
<tr>
<td>Patient3</td>
<td>40 years</td>
<td>Normal</td>
<td>Normal</td>
<td>none</td>
<td>Lip repair</td>
</tr>
</tbody>
</table>
DISCUSSION

The central part of the face and forebrain is closely related in the early phase of development. Embryologically, these deformities are due to defects in the development of the median part of the anterior neural plate. In the third week of gestation, the primitive tissues of trilaminar embryo give rise to the notochord and precordal mesoderm. At the same time, rostral ectoderm differentiates to form highly specialized neural crest cells. These tissues are responsible for the ultimate development of the brain and midline facial structures. Precordal mesoderm has a dual role. Its first role is in formation of the median skeleton of the face and the second role is in the induction and differentiation of rostral neural ectoderm and thereby prosencephalic development. Therefore, whenever prechordal mesoderm is defective mid facial and prosencephalic development may be arrested at any stage. During this stage, a developmental error results in holoprosencephaly and mid facial defects. Between the fourth and eight weeks rapid longitudinal growth of the embryo leads to the formation of five primary brain vesicles, which after further division and cleavage, form the cerebral hemisphere, lateral ventricles and olfactory and optic bulbs. Concurrent with the development of the brain the frontonasal prominence migrates to interpose between maxillary prominences. This central prominence is the precursors of midline structures including crista galli, ethmoid, vomer, cartilaginous nasal septum, premaxillary bones, maxillary incisors and soft tissue of forehead nose and prolabium. These deformities can be explained by defective development in these concurrent embryological events.

Within the spectrum of holoprosencephaly, the severity of the facial abnormality generally parallels with that of the brain. Grossly abnormal facies as seen in cyclopia, ethmocephaly or cebocephaly are associated with an alobar brain. Isolated cases of median cleft lip and hypotelorbitism with normal brain development have been reported. The craniofacial defects are centered around the ethmoid bone. Varying degree of hypoplasia with absence of cribiform and perpendicular plate is responsible for the failure of orbital separation abnormal migration of embryonic tissue into this area produces the proboscis seen in severe cases.

There are two main varieties of median facial anomalies. Hyperteloristic type or true median cleft occurs in varying degree from a notch in the vermilion to a complete cleft face. In the severe form hypertelorism, a bifid nose is present and rarely, it is associated with cleft palate or coloboma of the nasal ala. It results from lacking or incomplete fusion of the two medial nasal swellings in the midline. In the severe form the migration of all the bilateral facial structures forward and medially is inhibited. This occurs late in the organogenesis. The second type is hypoteloristic type or median pseudocleft with absent premaxilla, prolabium, columella, cleft palate, hypotelorism and holoprosencephaly. It is caused by disturbed development of prosencephalon to telencephalon (fore brain hemisphere) and diencephalons (optic vesicles) and incomplete division of both hemispheres. The result is holoprosencephaly and inhibited development of frontonasal process in to medial nasal swelling and to premaxilla prolabium and columella. This is a very early disturbance.

Rarity of cases, use of dubious terminology to describe the wide spectrum of clinical presentation of median facial malformation leads to confusion. In 1892, Kundrat used the term arhinencephaly to describe the absence of olfactory bulbs and tracts. The term holoprosencephaly was used by DeMayer and Zeman to describe the arrest in cleavage of prosencephalon in to cerebral hemisphere. These terms however, only refer to the cerebral anomalies and do not address the facial anomalies, which are invariably present. DeMayer et al classified holoprosencephaly into the following 5 groups.

1. Cyclopia – Only one eye develops because of fusion of the orbits and instead of a true nose, a proboscis is often located above the single orbit. In addition, there is hypoplasia or absence of many of the facial bones, low set ears, and an absent philtrum.

2. Ethmocephaly – There is hypotelorism and instead of a nose, a proboscis is usually located between the orbits. In addition, the ears are usually low set and there is an absent philtrum.

3. Cebocephaly – There is a rudimentary nose with
single nostril and hypotelorism. The ears are usually low set and the philtrum is again absent.

4. False median cleft lip – The cleft lip is directly in the midline and involves the area of the philtrum that is absent.

5. Median philtrum-premaxilla anlage

Holoprosencephaly is present in alobar form in group 1-4 and semi lobar or lobar in group 5. Gruss and Mathew later added one more group (group-6) unilateral cleft of primary palate with prolabium-premaxilla anlage to this classification and called the whole group as median cerebro-facial dysgenesis. Noordhoff and Cheng further modified this classification and coined the term median facial dysgenesis. This term was later changed to median facial dysplasia because dysplasia means developmental malformation while dysgenesis suggests abnormal morphogenesis. Ben- Hur et al have classified these patients in to a different subgroup called as median cleft lip with hypotelorism without holoprosencephaly. Van der Meulen classified these malformations based on embryology and the region involved. He recommended that the term dysgenesis should be changed to dysplasia and the terms median and cerebro-facial were appropriate for regional orientation.

The association of median brain and facial malformations led to the concept that “face predicts the brain.” According to DeMayer’s classification there is strong correlation between face and brain from group 1 to group 4; but the developmental journey from group 5 to a normal face is short and here correlation between face and brain weakened. Therefore saying that the face predicts the brain in cases of holoprosencephaly is correct only in 80% cases. However, the rest 20% cases are usually those who referred for evaluation and treatment.

Problems related to growth in cleft lip and palate can be better understood in these patients. Studies of patients with untreated cleft lip and palate revealed relatively normal growth patterns while un-operated MFD patients demonstrated deficient maxillary growth and all had significant growth disturbances following surgery. Therefore, surgery alone cannot be implicated as the sole cause for growth disturbance in these patients. The usual type of cleft lip and palate patients might be able to overcome the stress of surgery and have relatively normal growth. However patients with MFD with an inherently poor growth potential are unable to overcome the additional stress of surgery.

Between groups 5 to normal face various sub-groups of patients exist, who do not fit into any of the prevailing classifications. Such patients have pseudo median cleft lip and mid facial structures hypoplasia with hypotelorism without holoprosencephaly, unilateral cleft lip with hypotelorism and holoprosencephaly, midface hypoplasia with hypotelorism without false median cleft lip or median pseudo cleft lip without hypotelorism and without holoprosencephaly which was seen in our patients. These patients have normally developed brains, normal intelligence and potential for full life expectancy. Inclusion of these patients as holoprosencephaly disorders and their distinction from true median clefts are based on their embryological origin. All have in common the absolute deficit of tissue derived from frontonasal prominence. The presence of developmental delays and subtle neurological deficit in some patients without demonstrable brain abnormalities raises the possibilities of missing a link in the classification.

At the one end of spectrum, all the cases of severe malformations of the brain (De Mayer’s group 1-4) are incompatible with life and at the other end there are some patients with midline facial defects and normal or near normal brain development (De Mayer’s group 5 and intermediate groups). These patients have potential for full life expectancy and are candidate for surgical correction. Deficient horizontal growth of the maxilla produces characteristic dish face deformity that leads to severe psychological and functional problems. Surgical correction requires multidisciplinary approach and it needs to be tailored individually depending upon the degree of deformity. Before any treatment is instituted, all the specialists should evaluate these patients and formulate treatment plan.

Surgical closure of lip defect enhances the facial appearance and increase social acceptance. Basic
strategy for management of these patients remains the same but several alterations are necessary during primary nasolabial repair because of the diminutive bony and soft-tissue elements. Millard advocated the use of Abbe flap to close upper lip defect. Various bone and cartilage grafts are required to augment the nasal tip, columella and premaxillary region. Soft tissue augmentation in the form of dermal grafting to the median tubercle, philtral ridge, and basal columella may be required. Correction of skeletal defect will require for the hypotelorbitism, mid face retrusion, transverse maxillary arch deficiency and for nasal hypoplasia. All adolescent patients will need maxillary advancement and construction of nasal framework with costochondral or cranial graft.

Mild malocclusion can be corrected satisfactorily by orthodontic treatment while severe malocclusion particularly type III maxillary retrusion will require Le Fort I maxillary advancement.

Other problems faced in the management of patients with a lobar and semilobar holoprosencephaly are convulsions, difficulty with feeding, endocrine deficiency, electrolyte imbalance and temperature control. The prognosis for alobar holoprosencephaly is invariably poor. If not delivered as a stillbirth, these children usually die within the first year of life. Semi lobar holoprosencephaly, also have the same fate. Some cases have been reported to live into childhood. However, they have profound mental retardation. Children with lobar holoprosencephaly often survive childhood but still have some level of mental retardation. It is recommended that a genetic karyotype should be performed to help in defining the recurrence risk and genetic counseling of the parents is done.

REFERENCES